



## MEGF8 gene

multiple EGF like domains 8

### Normal Function

The *MEGF8* gene provides instructions for making a protein whose function is unclear. Based on its structure, the Megf8 protein may be involved in cell processes such as attaching cells to one another (cell adhesion) and helping proteins interact with each other. Researchers also suspect that the Megf8 protein plays a role in the normal shaping (patterning) of many parts of the body during embryonic development.

### Health Conditions Related to Genetic Changes

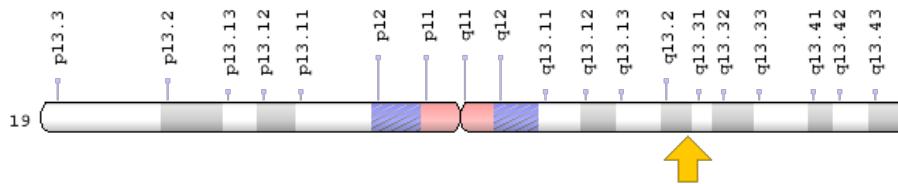
#### Carpenter syndrome

At least six mutations in the *MEGF8* gene have been found to cause Carpenter syndrome, a condition characterized by irregular skull formation, finger and toe abnormalities, and many other features. These mutations reduce or eliminate the function of the Megf8 protein. Researchers suspect that the amount of protein function that is retained may contribute to the variability in signs and symptoms. It is unclear how *MEGF8* gene mutations cause Carpenter syndrome. The mutations likely interfere with normal patterning of many parts of the body, which contributes to the features of this disorder.

### Chromosomal Location

Cytogenetic Location: 19q13.2, which is the long (q) arm of chromosome 19 at position 13.2

Molecular Location: base pairs 42,325,609 to 42,378,769 on chromosome 19 (Homo sapiens Annotation Release 108, GRCh38.p7) (NCBI)



Credit: Genome Decoration Page/NCBI

## **Other Names for This Gene**

- C19orf49
- EGF-like domain-containing protein 4
- EGF-like-domain, multiple 4
- EGFL4
- epidermal growth factor-like protein 4
- FLJ22365
- HBV pre-s2 binding protein 1
- MEGF8\_HUMAN
- multiple EGF-like-domains 8
- multiple epidermal growth factor-like domains protein 8
- SBP1

## **Additional Information & Resources**

### Scientific Articles on PubMed

- PubMed  
<https://www.ncbi.nlm.nih.gov/pubmed?term=%28MEGF8%5BTIAB%5D%29+AND+%28%28Genes%5BMH%5D%29+OR+%28Genetic+Phenomena%5BMH%5D%29%29+AND+english%5Bla%5D+AND+human%5Bmh%5D+AND+%22last+3600+days%22%5Bdp%5D>

### OMIM

- MULTIPLE EPIDERMAL GROWTH FACTOR-LIKE DOMAINS 8  
<http://omim.org/entry/604267>

### Research Resources

- Atlas of Genetics and Cytogenetics in Oncology and Haematology  
[http://atlasgeneticsoncology.org/Genes/GC\\_MEGF8.html](http://atlasgeneticsoncology.org/Genes/GC_MEGF8.html)
- ClinVar  
<https://www.ncbi.nlm.nih.gov/clinvar?term=MEGF8%5Bgene%5D>
- HGNC Gene Symbol Report  
[http://www.genenames.org/cgi-bin/gene\\_symbol\\_report?q=data/hgnc\\_data.php&hgnc\\_id=3233](http://www.genenames.org/cgi-bin/gene_symbol_report?q=data/hgnc_data.php&hgnc_id=3233)

- NCBI Gene  
<https://www.ncbi.nlm.nih.gov/gene/1954>
- UniProt  
<http://www.uniprot.org/uniprot/Q7Z7M0>

## Sources for This Summary

- OMIM: MULTIPLE EPIDERMAL GROWTH FACTOR-LIKE DOMAINS 8  
<http://omim.org/entry/604267>
- Perlyn CA, Marsh JL. Craniofacial dysmorphology of Carpenter syndrome: lessons from three affected siblings. *Plast Reconstr Surg.* 2008 Mar;121(3):971-81. doi: 10.1097/01.prs.0000299284.92862.6c.  
*Citation on PubMed:* <https://www.ncbi.nlm.nih.gov/pubmed/18317146>
- Twigg SR, Lloyd D, Jenkins D, Elçioglu NE, Cooper CD, Al-Sanna N, Annagür A, Gillessen-Kaesbach G, Hüning I, Knight SJ, Goodship JA, Keavney BD, Beales PL, Gileadi O, McGowan SJ, Wilkie AO. Mutations in multidomain protein MEGF8 identify a Carpenter syndrome subtype associated with defective lateralization. *Am J Hum Genet.* 2012 Nov 2;91(5):897-905. doi: 10.1016/j.ajhg.2012.08.027. Epub 2012 Oct 11.  
*Citation on PubMed:* <https://www.ncbi.nlm.nih.gov/pubmed/23063620>  
*Free article on PubMed Central:* <https://www.ncbi.nlm.nih.gov/pmc/articles/PMC3487118/>
- Zhang Z, Alpert D, Francis R, Chatterjee B, Yu Q, Tansey T, Sabol SL, Cui C, Bai Y, Koriabine M, Yoshinaga Y, Cheng JF, Chen F, Martin J, Schackwitz W, Gunn TM, Kramer KL, De Jong PJ, Pennacchio LA, Lo CW. Massively parallel sequencing identifies the gene Megf8 with ENU-induced mutation causing heterotaxy. *Proc Natl Acad Sci U S A.* 2009 Mar 3;106(9):3219-24. doi: 10.1073/pnas.0813400106. Epub 2009 Feb 13.  
*Citation on PubMed:* <https://www.ncbi.nlm.nih.gov/pubmed/19218456>  
*Free article on PubMed Central:* <https://www.ncbi.nlm.nih.gov/pmc/articles/PMC2651267/>

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Reprinted from Genetics Home Reference:

<https://ghr.nlm.nih.gov/gene/MEGF8>

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